Single Nucleotide Polymorphism

Reference SNP(refSNP) Cluster Report: rs2066844

BUILD 129
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Go

refSNP ID: rs2066844
Organism: human (*Homo sapiens*)
Molecule Type: Genomic
Created/Updated in build: 94/129
Map to Genome Build: 36.3
Citation: PubMed

Allele
Variation Class: SNP: single nucleotide polymorphism
RefSNP Alleles: C/T
Ancestral Allele: Not available
Clinical Association: unknown

HGVS Names Links | Lineage
NM_022162.1:c.2104C>T
NP_071445.1:p.R702W
NT_010498.15:g.4360124C>T

GENERAL
HUMAN VARIATION
Search, Annotate, Submit NEW
Annotate and Submit Batch Data with Clinical Impact NEW
SNP SUBMISSION
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SNP Details are organized in the following sections:

Submission Fasta Resource GeneView Map Diversity Validation

Submitter records for this RefSNP Cluster

The submission **ss48412844** has the longest flanking sequence of all cluster members and was used to instantiate sequence for **rs2066844** during BLAST analysis for the current build.

NCBI Assay ID	Handle|Submitter ID	Validation Status	ss to rs Orientation /Strand	Alleles	5' Near Seq 30 bp	3' Near Seq 30 bp	Entry Date	Update Date	Build Added	Mo T
ss2978536	CEPH|IBD1-SNP8	K	fwd/B	C/T	gagtgccagacatctgagaaggcctgctc	ggcgccaggcctgtgccccgctggtgtctgg	03/22/01	10/25/06	94	Ger
ss2992222	GKT-CGM|SNP-EX4.4	K	fwd/B	C/T	gagtgccagacatctgagaaggcctgctc	ggcgccaggcctgtgccccgctggtgtctgg	05/30/01	10/25/06	96	Ger
ss7987100	HPGA-WEISS-MARTINEZ|HPGA-CARD15_17379		fwd/B	C/T	gagtgccagacatctgagaaggcctgctc	ggngccaggcctgtgccccgctggtgtctgg	04/08/03	10/10/03	114	Ger
ss8819603	SNP500CANCER|CARD15-02	K	fwd/B	C/T	gagtgccagacatctgagaaggcctgctc	ggcgccaggcctgtgccccgctggtgtctgg	05/30/03	04/07/04	116	Ger
ss24523902	PERLEGEN|afd4338565		fwd/B	C/T	gagtgccagacatctgagaaggcctgctc	ggcgccaggcctgtgccccgctggtgtctgg	08/10/04	08/21/04	123	Ger
ss28514849	JDRF_WT_DIL|DIL2226		rev/T	A/G	ccagacaccagcgggcacaggcctggcgcc	gagcagggcctctcagatgtctggcactc	09/07/04	09/07/04	126	Ger
ss48412844	APPLERA_GI|hCV11717468	K	fwd/B	C/T	gagtgccagacatctgagaaggcctgctc	ggcgccaggcctgtgccccgctggtgtctgg	09/28/05	11/03/06	126	Ger
ss74879819	ILLUMINA|ILMN_Human_1M_rs2066844		fwd/B	C/T	gagtgccagacatctgagaaggcctgctc	ggcgccaggcctgtgccccgctggtgtctgg	08/28/07	08/29/07	129	Ger
ss84172810	PHARMGKB_CREATE|PS204942_PA141943057_87		fwd/B	C/T	gagtgccagacatctgagaaggcctgctc	ggcgccaggcctgtgccccgctggtgtctgg	12/06/07	12/10/07	130	Ger
ss86342483	CANCER-GENOME|10586		fwd/B	C/T	gagtgccagacatctgagaaggcctgctc	ggcgccaggcctgtgccccgctggtgtctgg	01/25/08	01/25/08	129	Ger

Fasta sequence (Legend)

>gnl|dbSNP|rs2066844|allelePos=301|totalLen=601|taxid=9606|snpclass=1|alleles='C/T'|mol=Genomic|build=130

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CCACCAGCTT TGCTCAGACA CCTCTTCAAT TGTGGCAGGC CAGGCAACTC ACCAATGGCC
AGGCTCCTGC CCACGATGTG CATCCAGGCC TCGGAGGGAA AGGACAGCAG CGTGGCAGCT
TTGCTGCAGA AGGCCGAGCC GCACAACCTT CAGATCACAG CAGCCTTCCT GGCAGGGCTG
TTGTCCCGGG AGCACTGGGG CCTGCTGGCT GAGTGCCAGA CATCTGAGAA GGCCCTGCTC
Y
GGCGCCAGGC CTGTGCCCGC TGGTGTCTGG CCCGCAGCCT CCGCAAGCAC TTCCACTCCA
TCCCGCCAGC TGCACCCGGT GAGCCAAGA GCGTGCATGC CATGCCCGGG TTCATCTGGC
TCATCCGGAG CCTGTACGAG ATGCAGGAGG AGCGGCTGGC TCGGAAGGCT GCACGTGGCC
TGAATGTTGG GCACCTCAAG TTGACATTTT GCAGTGTGGG CCCCACTGAG TGTGCTGCCC
TGGCCTTTGT GCTGCAGCAC CTCCGGCGGC CCGTGGCCCT GCAGCTGGAC TACAACTCTG
```

GeneView

GeneView via analysis of contig annotation: NOD2 nucleotide-binding oligomerization domain containing 2

View variations for gene: Include clinically associated: ○ in gene region ○ cSNP ○ has frequency ○ double hit Go

Group Label	Contig->mRNA	Gene Model (contig mRNA transcript) Color Legend
reference	NT_010498->NM_022162	
	function	

Group label	Contig->mRNA->Protein	Contig position	mRNA orientation	mRNA pos	Function	dbSNP allele	Protein residue	Codon pos	Amino acid pos
reference	NT_010498->NM_022162->NP_071445	4360125	forward	2209	missense	T	Trp [W]	1	702
					contig reference	C	Arg [R]	1	702

GeneView: no link established by BLAST analysis of mRNA sequences

Integrated Maps:

NCBI MapViewer: rs2066844 maps exactly once on NCBI human chromosome 16

Chromosome	Contig accession	Contig position	Chromosome position	Hit orientation	Contig Allele	Assembly Type	Group label	Contig label	Neighbor SNP	SNP_flank position
16	NW_926462.1	4326052	35261253	plus	C	alt_assembly_1	Celera	Celera	view	300
16	NW_001838288.2	527844	36632893	minus	G	alt_assembly_8	HuRef	HuRef	view	300
16	NT_010498.15	4360125	49303427	plus	C	ref_assembly	reference	reference	view	300

NCBI Resource Links

Submitter-Referenced		dbSNP Blast Analysis		UniGene Cluster ID
dbSTS	GenBank	NCBI RefSeq NM (mRNA):	GenBank mRNA:	135201
G67950	NT_019610.3 NM_022162.1	NM_022162.1	AF178930.1	

Population Diversity

ss#	Population	Individual Group	Chrom. Sample Cnt.	Source	Sample Ascertainment: Genotype Detail NEW C/C	C/T	HWP	Alleles: C	T
ss24523902	AFD_EUR_PANEL	European	48	IG	0.958	0.042	1.000	0.979	0.021
	AFD_AFR_PANEL	African American	46	IG	1.000			1.000	
	AFD_CHN_PANEL	Asian	48	IG	1.000			1.000	
ss2978536	EUCAUC		24	AF				0.830	0.170
ss2992222	CD_UK-POP		64	AF				0.860	0.140
ss48412844	HapMap-CEU	European	118	IG	0.780	0.220	0.343	0.890	0.110
	HapMap-HCB	Asian	90	IG	1.000			1.000	
	HapMap-JPT	Asian	90	IG	1.000			1.000	
	HapMap-YRI	Sub-Saharan African	118	IG	1.000			1.000	
	AGI_ASP population	multiple	78	IG	0.949	0.051	1.000	0.974	0.026
ss7987100	D-0	African American	48	IG	1.000			1.000	
	E-0	European	40	IG	0.950	0.050	1.000	0.975	0.025

	E-1	European	6	IG	1.000	1.000
ss8819693	P1		204	GF	0.951 0.049 1.000	0.975 0.025
	CAUC1		62	GF	0.871 0.129 0.752	0.936 0.065
	AFR1		48	GF	1.000	1.000
	HISP1		46	GF	0.957 0.043 1.000	0.979 0.022
	PAC1		48	GF	1.000	1.000

Summary	Average Het.+/- std err:	Individual Count	Founders Count	Individual Overlap	Genotype Conflict
	0.046	372	300	46	0

Validation Summary:

Validation status	Marker displays Mendelian segregation	PCR results confirmed in multiple reactions	Homozygotes detected in individual genotype data
	YES	YES	YES

GENERAL: Contact Us | Homepage | Announcements | dbSNP Summary | Genome | FTP SERVER | Build History | Handle Request
DOCUMENTATION: FAQ | Searchable FAQ Archive | Overview | How to Submit | RefSNP Summary Info | Database Schema
SEARCH: Entrez SNP | Blast SNP | Batch Query | By Submitter | New Batches | Method | Population | Publication | Batch | Locus Info | Between Marker
HAPLOTYPE: Submission | Specifications | Sample HapSet | Sample Individual
NCBI: PubMed | Entrez | BLAST | OMIM | Taxonomy | Structure

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Single Nucleotide Polymorphism

Reference SNP(refSNP) Cluster Report: rs2066845

BUILD 129
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Annotate and Submit Batch Data with Clinical Impact NEW
SNP SUBMISSION
DOCUMENTATION
SEARCH
RELATED SITES

refSNP ID: rs2066845
Organism: human (*Homo sapiens*)
Molecule Type: Genomic
Created/Updated in build: 94/129
Map to Genome Build: 36.3
Citation: PubMed

Allele	HGVS Names (Links | Lineage)
Variation Class: SNP: single nucleotide polymorphism	NM_022162.1:c.2722G>C ←
RefSNP Alleles: C/G	NP_071445.1:p.G908R
Ancestral Allele: Not available	NT_010498.15:g.4370738G>C
Clinical Association: unknown	

SNP Details are organized in the following sections:

Submission Fasta Resource GeneView Map Diversity Validation

Submitter records for this RefSNP Cluster

The submission **ss48412842** has the longest flanking sequence of all cluster members and was used to instantiate sequence for **rs2066845** during BLAST analysis for the current build.

NCBI Assay ID	Handle|Submitter ID	Validation Status	ss to rs Orientation /Strand	Alleles	5' Near Seq 30 bp	3' Near Seq 30 bp	Entry Date	Update Date	Build Added	Mo T
ss2978537	CEPH|BD1-SNP12		fwd/B	C/G	gttgactctttggcctttcagattc:tgg	gcaacagagtgggtgacgaggggcccagg	03/22/01	04/07/04	94	Ger
ss2992223	GKT-CGM|SNP-EX8.1		fwd/B	C/G	gttgactctttggcctttcagattc:tgg	gcaacagagtgggtgacgaggggcccagg	05/30/01	10/25/06	96	Ger
ss12675296	SNP500CANCERCARD15-03		fwd/B	C/G	gttgactctttggcctttcagattc:tgg	gcaacagagtgggtgacgaggggcccagg	09/05/03	04/07/04	118	Ger
ss24524028	PERLEGEN|af4228335		fwd/B	C/G	gttgactctttggcctttcagattc:tgg	gcaacagagtgggtgacgaggggcccagg	08/10/04	09/13/04	123	Ger
ss28514841	JDRF_WT_DIL|DIL2227		rev/T	C/G	cctgggcccctcgtcacccactctgctgc	ccagaatctgaaaaggccaaaagagtcaac	09/07/04	09/07/04	126	Ger
ss48412842	APPLERA_GI|hCV11717466		fwd/	C/G	gttgactctttggcctttcagattctgg	gcaacagagtgggtgacgaggggcccagg	09/28/05	11/03/06	126	Ger
ss74807768	AFFY|SNP_M-178946		fwd/B	C/G	gttgactctttggcctttcagattctgg	gcaacagagtgggtgacgaggggcccagg	08/09/07	08/09/07	128	Ger
ss84172775	PHARMGKB_CREATE|PS204943_PA141942202_99		fwd/	C/G	gttgactctttggcctttcagattctgg	gcaacagagtgggtgacgaggggcccagg	12/06/07	12/10/07	130	Ger
ss84172805	PHARMGKB_CREATE|PS204942_PA141942769_99		fwd/	C/G	gttgactctttggcctttcagattctgg	gcaacagagtgggtgacgaggggcccagg	12/06/07	12/10/07	130	Ger
ss86342486	CANCER-GENOME|7917		fwd/	C/G	gttgactctttggcctttcagattctgg	gcaacagagtgggtgacgaggggcccagg	01/25/08	01/25/08	129	Ger

Fasta sequence (Legend)

>gnl|dbSNP|rs2066845|allelePos=301|totalLen=601|taxid=9606|snpclass=1|alleles='C/G'|mol=Genomic|build=130

```
CTCTTGTCAG TGAGTTCCTG TCCTTAAGGG TTAGGGCTGG GTAGCCCTCT ACTATTCTCT
AAGTCTGTAA TGTAAAGCCA CTGAAAACTC TTGGGTTAAG TTTGGCCATC CCACCCAAAA
GATGGAGGCA GGTCCACTTT GCTGGGACCA GGAGCCCCAG TGAGGCCACT CTGGGATTGA
GTGGTCCTGC CCCTCTGGCT GGGACTGCAG AGGGAGGAGG ACTGTTAGTT CATGTCTAGA
ACACATATCA GGTACTCACT GACACTGTCT GTTGACTCTT TTGGCCTTTT CAGATTCTGG
G
GCAACAGAGT GGGTGACGAG GGGGCCCAGG CCCTGGCTGA AGCCTTGGGT GATCACCAGA
GCTTGAGGTG GCTCAGGTAA GCTTCAGAGT CTATCCTGCA GTTTTCTTGG GGAGATCAGG
TGAAGAGGGA GGAGCTGGGG CCAGTTCTGA AGGTCTTTGA ACTTTATTTC TACCCCACAA
TGTTAGGCAA TGGAGTAAGG AAAAAAGACC ATTGGATTTC AAGAGAGGAC ACTCGAGTCT
TTCTGGGTGA CTTGGAAATG TCCCTTGTCC TCTCAGGGTT TTGATACAGT ATCTGTAAAT
```

GeneView

GeneView via analysis of contig annotation: NOD2 nucleotide-binding oligomerization domain containing 2

View variations for gene: Include clinically associated: in gene region cSNP has frequency double hit Go

Group Label	Contig->mRNA	Gene Model (contig mRNA transcript) Color Legend
reference	NT_010498->NM_022162	
	function	

Group label	Contig->mRNA->Protein	Contig position	mRNA orientation	mRNA pos	Function	dbSNP allele	Protein residue	Codon pos	Amino acid pos
reference	NT_010498->NM_022162->NP_071445	4370739	forward	2827	missense	C	Arg [R]	1	908
					contig reference	G	Gly [G]	1	908

GeneView: no link established by BLAST analysis of mRNA sequences

Integrated Maps:

NCBI MapViewer: rs2066845 maps exactly once on NCBI human chromosome 16

Chromosome	Contig accession	Contig position	Chromosome position	Hit orientation	Contig Allele	Assembly Type	Group label	Contig label	Neighbor SNP	SNP_flank position
16	NW_926462.1	4336666	35271867	plus	G	alt_assembly_1	Celera	Celera	view	300
16	NW_001838288.2	517233	36643504	minus	C	alt_assembly_8	HuRef	HuRef	view	300
16	NT_010498.15	4370739	49314041	plus	G	ref_assembly	reference	reference	view	300

NCBI Resource Links

Submitter-Referenced	dbSNP Blast Analysis	UniGene Cluster ID
dbSTS GenBank	NCBI RefSeq NM (mRNA):	135201
G67951 NT_019610.3 NM_022162.1	NM_022162.1	

Population Diversity

ss#	Population	Individual Group	Chrom. Sample Cnt.	Source	Sample Ascertainment: Genotype Detail NEW C/G	G/G	HWP	Alleles: C	G
ss12675296	PI		204	GF	0.010	0.990	1.000	0.005	0.995
	CAUC1		62	GF		1.000			1.000
	AFR1		48	GF		1.000			1.000
	HISP1		46	GF		1.000			1.000
	PAC1		48	GF	0.042	0.958	1.000	0.021	0.979
ss24524028	AFD_EUR_PANEL	European	46	IG	0.087	0.913	1.000	0.043	0.957
	AFD_AFR_PANEL	African American	46	IG		1.000			1.000
	AFD_CHN_PANEL	Asian	48	IG		1.000			1.000
ss2978537	EUCAUC		20	AF				0.150	0.850
	CEPH		184	AF					1.000
ss2992223	CD_UK-POP		64	AF				0.060	0.940
ss48412842	HapMap-CEU	European	120	IG	0.033	0.967	1.000	0.017	0.983

HapMap-HCB	Asian	90	IG	1.000			1.000	
HapMap-JPT	Asian	88	IG	1.000			1.000	
HapMap-YRI	Sub-Saharan African	118	IG	1.000			1.000	
AGI_ASP population	multiple	78	IG	0.026	0.974	1.000	0.013	0.987

Summary	Average Het.+/- std err:	Individual Count	Founders Count	Individual Overlap	Genotype Conflict
	0.013 0.000	371	299	9	0

Validation Summary:

Validation status	Marker displays Mendelian segregation	PCR results confirmed in multiple reactions	Homozygotes detected in individual genotype data
	YES	YES	YES

Revised: May 25, 2006 1:38 PM .

Reference SNP(refSNP) Cluster Report: rs2066847

refSNP ID: rs2066847
Organism: human (*Homo sapiens*)
Molecule Type: Genomic
Created/Updated in build: 94/129
Map to Genome Build: 36.3
Citation: NHGRI GWAS PubMed

Allele
Variation Class: DIP: deletion/insertion polymorphism
RefSNP Alleles: -/C
Ancestral Allele: Not available
Clinical Association: unknown

HGVS Names Links
NM_022162.1:c.3016_3017insC
NT_010498.15:g.4377977_4377978insC

SNP Details are organized in the following sections:
Submission Fasta Resource GeneView Map Diversity Validation

Submitter records for this RefSNP Cluster

The submission **ss8819692** has the longest flanking sequence of all cluster members and was used to instantiate sequence for **rs2066847** during BLAST analysis for the current b

NCBI Assay ID	Handle|Submitter ID	Validation Status	ss to rs Orientation /Strand	Alleles	5' Near Seq 30 bp	3' Near Seq 30 bp	Entry Date	Update Date	Build Added	Molecule Type	Freq Warni
ss2978539	CEPHIBD1-SNP13		fwd/T	-/C	cctacctaggggcagaagcoctcctgcagg	ccttgaaaggaatgacacacatcctggaag	03/22/01	10/25/06	94	Genomic	
ss2992224	GKT-CGMISNP-EX11.1/ins		fwd/T	-/C	cctacctaggggcagaagcoctcctgcagg	ccttgaaaggaatgacacacatcctggaag	05/30/01	10/25/06	96	Genomic	
ss8819692	SNP500CANCERCARD15-01		fwd/T	-/C	cctacctaggggcagaagcoctcctgcagg	ccttgaaaggaatgacacacatcctggaag	05/30/03	04/07/04	116	Genomic	

Fasta sequence (Legend)

>gnl|dbSNP|rs2066847|allelePos=142|totalLen=330|taxid=9606|snpclass=2|alleles='-/C'|mol=Genomic|build=116

GACTGGCTAA CTCCTGCAGT CTCTTTAACT GGACAGTTTC AAGAGGAAAA CCAAGAATCC
TTGAAGCTCA CCATTGTATC TTCTTTTCCA GGTTGTCCAA TAACTGCATC ACCTACCTAG
GGGCAGAAGC CCTCCTGCAG G
N
CCCTTGAAAG GAATGACACC ATCCTGGAAG TCTGGTAAGG Ccoctgggca ggcctgtttt
agctctccga acctcagttt ttctatctgt aaaatggggt gacgggagag aggaatggca
gaattttgag gatcccttct gattctgaca ttcagtgaGA ATGATTCTGC ATGTGAAGGA
TCTGATTC

GeneView

GeneView via analysis of contig annotation: NOD2 nucleotide-binding oligomerization domain containing 2

View variations for gene: Include clinically associated: in gene region cSNP has frequency double hit Go

Group Label	Contig->mRNA	Gene Model (contig mRNA transcript) Color Legend
reference	NT_010498->NM_022162	

function

Group label	Contig-->mRNA-->Protein	Contig position	mRNA orientation	mRNA pos	Function	dbSNP allele	Protein residue	Protein pos	Codon pos	Amino acid
reference	NT_010498->NM_022162->NP_071445	4377977:4377978	forward	3121	frame shift	C	Pro [P]	1	1006	
					contig reference	-		1	1006	

GeneView: no link established by BLAST analysis of mRNA sequences

Integrated Maps:

NCBI MapViewer: rs2066847 maps exactly once on NCBI human chromosome 16

Chromosome	Contig accession	Contig position	Chromosome position	Hit orientation	Contig Allele	Assembly Type	Group label	Contig label	Neighbor SNP	SNP_flank position
16	NW_926462.1	4343903^4343904	35279104^35279105	plus	-	alt_assembly_1	Celera	Celera	view	141..141
16	NW_001838288.2	509995^509996	36650742^36650743	minus	-	alt_assembly_8	HuRef	HuRef	view	141..141
16	NT_010498.15	4377977^4377978	49321279^49321280	plus	-	ref_assembly	reference	reference	view	141..141

NCBI Resource Links

Submitter-Referenced		dbSNP Blast Analysis	UniGene Cluster ID
dbSTS	GenBank		135201
G67955	NT_019610.3 NM_022162.1		

Population Diversity

ss#	Population	Sample Ascertainment: Individual Group	Chrom. Sample Cnt.	Source	Genotype Detail NEW: +/-	-/-	HWP	Alleles: +	-	C
ss2978539	EUCAUC		20	AF					0.700	0.300
ss2992224	CD_UK-POP		64	AF					0.890	0.110
ss8819692	P1		200	GF	0.010	0.990	1.000	0.005	0.995	
	CAUC1		62	GF		1.000			1.000	
	AFR1		48	GF		1.000			1.000	
	HISP1		44	GF	0.045	0.955	1.000	0.023	0.978	
	PAC1		46	GF		1.000			1.000	

Summary	Average Het.+/- std err:	Individual Count	Founders Count	Individual Overlap	Genotype Conflict
	0.010 +/- 0.000	0	0	0	0

Validation Summary:

Validation status	Marker displays Mendelian segregation	PCR results confirmed in multiple reactions	Homozygotes detected in individual genotype data

YES	YES	YES

GENERAL: Contact Us | Homepage | Announcements |dbSNP Summary | Genome | FTP SERVER | Build History | Handle Request
DOCUMENTATION: FAQ | Searchable FAQ Archive | Overview | How to Submit | RefSNP Summary Info | Database Schema
SEARCH: Entrez SNP | Blast SNP | Batch Query | By Submitter |New Batches | Method | Population | Publication | Batch | Locus Info | Between Marker
HAPLOTYPE:Submission | Specifications | Sample HapSet | Sample Individual
NCBI: PubMed | Entrez | BLAST | OMIM | Taxonomy | Structure

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